National Registry of Genetically Triggered Thoracic Aortic Aneurysms and Cardiovascular Conditions (GenTAC)

August 2013

GenTAC Clinical Center Highlight: University of Texas Medical School at Houston

In this issue we highlight the University of Texas Medical School at Houston (UT Health), where Dianna Milewicz MD, PhD is the Principal Investigator. Dr. Milewicz is professor and director of the Division of Medical Genetics in the Department of Internal Medicine at UT Health and serves as the principal investigator for the UT Health and Texas Children's Hospital. She is director of the MD/PhD Program, a joint program between UT and the M.D. Anderson Cancer Center, as well as Director of the John Ritter Research Program in Aortic and Vascular Diseases with the University of Texas Health Science Center. Her co-Investigator for GenTAC is Dr. Siddharth Prakash, MD, PhD, F.A.C.C., F.A.H.A. He is Assistant Professor in the Department of Internal Medicine. The GenTAC study coordinator is Meghan Terry who has worked in research for 8 years, including in pharmaceutical trials, before joining the GenTAC team.

GenTAC Steering Committee Meeting, Rockville MD

Recent Highlights

- On April 15, 2013, GenTAC held its annual Steering Committee meeting in Rockville MD. Several guest speakers presented their research including Alan Braverman (Washington University) and his findings on in Women with Aortopathy, and Sherene Shalhub (University of Texas, Houston) and her research on Ehlers Danlos. We also had the opportunity to recognize Carolyn Bondy for her lifetime contributions to improve the lives of patients with Turner syndrome.
- Videocasts of the GenTAC investigators discussing the research they have done using the GenTAC Registry are available on the GenTAC website. Currently you can see Drs. H. Eser Tolunay and Patrice Desvigne-Nickens, introduce and discuss the origins of the Registry. Click here to view. Check back regularly as new videocasts will be posted to the site.

GenTAC will be at AHA!

Please join us Tuesday, November 19 from 7:30-8:45am for this Scientific Session on ***Epidemiology and Genetics of Adult Congenital Heart Disease: Insights from GenTAC***, the National Registry of Genetically Trig gered Aneurysms and Other Cardiovascular Conditions.
Moderators: Kim A Eagle, Ann Arbor, MI; Cheryl L Maslen, Portland, OR

Presentations:

Tuesday, Nov 19, 2013, 7:30 AM - 7:45 AM
SNPs and rare copy number variants are associated with bicuspid aortic valve in Turner syndrome.
Siddharth Prakash, Houston, TX

Tuesday, Nov 19, 2013, 7:45 AM - 8:00 AM
Calcium Channel Blockers Impose a Deleterious Gene-by-Environment Interaction in Marfan Syndrome: Implications for Aortic Disease Pathogenesis and Treatmen
Jefferson J Doyle, Baltimore, MD

Tuesday, Nov 19, 2013, 8:00 AM - 8:15 AM
Utilization and outcomes of valve sparing aortic root replacement in patients with Marfan syndrome enrolled in GenTAC
Howard Song, Portland, OR

Tuesday, Nov 19, 2013, 8:15 AM - 8:30 AM
Gender differences among individuals with genetically-triggered thoracic aortic aneurysms and dissections
Kathryn W Holmes, Portland, OR

Tuesday, Nov 19, 2013, 8:30 AM - 8:45 AM
A unified protocol for analysis of the aorta results in high reproducibility of measurements across imaging modalities: the GenTAC iCORE experience
Federico M Asch, Washington, DC

Steering Committee Members

Principal Investigators	
Scott A. LeMaire, MD	Baylor College of Medicine
William Ravekes, MD	Johns Hopkins University School of Medicine
Nazli B. McDonnell, MD, PhD	NIA at Harbor Hospital
Cheryl L. Maslen, PhD	Oregon Health & Science University
Ralph V. Shohet, MD	Queen's Medical Center
Reed E. Pyeritz, MD, PhD	University of Pennsylvania School of Medicine
Dianna M. Milewicz, MD, PhD	University of Texas Medical School at Houston
Richard B. Devereux, MD	Weill Cornell Medical College of Cornell University
Core Labs	
Jennifer P. Habashi, MD	Johns Hopkins University Hospital
Federico M. Asch, MD	MedStar Research Institute
Data Coordinating Center	
Barbara L. Kroner, PhD	RTI International
NHLBI	
Eser H. Tolunay, PhD	National Heart, Lung, and Blood Institute
SC Chair	
Kim A. Eagle, MD	University of Michigan

Presented Abstracts

- Shalhub, Sherene. COL3A1 gene mutation predicts arterial involvement and prognosis in Vascular Ehlers Danlos Syndrome. Plenary Session. ***Society for Vascular Surgery,*** May 30–June 1 2013
- Asch, Federico. Are unicuspid and bicuspid aortic valve different phenotypes of the same disease? An insight from the GenTAC registry. ***American Society of Echocardiography***, June 29–July 2, 2013
- Asch, Federico. A unified protocol for analysis of the aorta results in high reproducibility of measurements across imaging modalities. The GenTAC registry. ***American Society of Echocardiography***, June 29–July 2, 2013

Save the date

GenTAC is hosting the Third GenTAC Thoracic Aortic Disease Summit in Baltimore, MD on July 9-10, 2014. It dovetails with the annual NMF meeting which is being held **July 10 – 13, 2014.** Information about the meeting will be on the GenTAC website as it becomes available.

Do you have a research interest in genetically triggered thoracic aortic conditions?

GenTAC makes its collection of medical data and biologic samples available at no cost to qualified investigators. Your work can help determine best practices that advance the clinical management of genetic aortic aneurysms and other cardiovascular conditions.

A Snapshot of Who is Enrolled in GenTAC

Number of people enrolled: 3546

Biospecimens

Blood:	2023
Saliva:	1223
Both blood and saliva	85
Tissue:	141

Gender

Male:	2110
Female:	1412

Eligible Diagnosis

Marfan:	870
Turner:	260
Ehlers-Danlos (vascular):	141
Ehlers-Danlos (other):	21
Loeys-Dietz:	99
FBN1, TGFBR mutation:	37
BAV with aortic enlargement:	883
BAV with family history:	25
BAV with coarctation:	78
Shprintzen-Goldberg:	5
Familial TAA:	267
Other aneurysm, dissections:	717
Other congenital heart disease:	94

Age

<5	113
5–7	607
18–39	1069
40–69	1621
>69	110

Race

White, non-Hispanic	2866
Black, non-Hispanic	180
Hispanic	276
Asian	132
American Indian/Alaskan native	17
Native Hawaiian/Pac. Islander	52

» To submit a proposal to use GenTAC data or for more information, visit our website: **http://gentac.rti.org.**